

WHAT IS CLAIMED IS:

- 1 1. An isolated nucleic acid comprising a nucleic acid sequence selected
2 from the group consisting of:
 - 3 a. nucleic acid sequences corresponding to the nucleic acid of
4 SEQ ID NO:1;
 - 5 b. nucleic acid sequences corresponding to the nucleic acid
6 sequences selected from the group consisting of SEQ ID NO:3, SEQ ID NO:5, SEQ ID
7 NO:7;
 - 8 c. nucleic acid sequences corresponding to the nucleic acid
9 sequence of SEQ ID NO:9; and
 - 10 d. nucleic acid sequences corresponding to the nucleic acid
11 sequences selected from the group consisting of SEQ ID NO:10, SEQ ID NO:11, and
12 SEQ ID NO:12.
- 1 2. The nucleic acid of claim 1, wherein the nucleic acid is genomic
2 DNA.
- 1 3. The nucleic acid of claim 2, wherein the DNA is cDNA.
- 1 4. The nucleic acid of claim 1, wherein the nucleic acid is a nucleic
2 acid sequence corresponding to the nucleic acid of SEQ ID NO:1.
- 1 5. The nucleic acid of claim 1, wherein the nucleic acid is a nucleic
2 acid sequence corresponding to the nucleic acid sequence of SEQ ID NO:9.
- 1 6. The nucleic acid of claim 1 wherein the nucleic acid is a nucleic
2 acid sequence corresponding to a nucleic acid sequence selected from the group consisting
3 of SEQ ID NO:3, SEQ ID NO:5, and SEQ ID NO:7.

1 7. The nucleic acid of claim 1, wherein the nucleic acid is a nucleic
2 acid sequence corresponding to a nucleic acid sequence selected from the group consisting
3 of SEQ ID NO:10, SEQ ID NO:11, and SEQ ID NO:12.

1 8. A nucleic acid comprising an RNA equivalent of the nucleic acid of
2 claim 1.

1 9. A cloning vector comprising a coding sequence of a nucleic acid as
2 set forth in any one of claims 1 through 7 and a replicon operative in a host cell for the
3 vector.

1 10. An expression vector comprising a coding sequence of a nucleic
2 acid set forth in any one of claims 1 through 7 operably linked with a promoter sequence
3 capable of directing expression of the coding sequence in host cells for the vector.

1 11. Host cells transformed with a vector as set forth in any one of
2 claims 9 and 10.

1 12. A method of producing a mutant HH polypeptide comprising:
2 a. transforming host cells with a vector capable of expressing a
3 polypeptide from a nucleic acid sequence as set forth in any one of claims 6 and 7;
4 b. culturing the cells under conditions suitable for production of
5 the polypeptide; and
6 c. recovering the polypeptide.

1 13. A peptide product selected from the group consisting of:
2 a. a polypeptide having the amino acid sequence corresponding
3 to the sequence of SEQ ID NO:2;
4 b. a polypeptide having the amino acid sequence corresponding
5 to the sequence of SEQ ID NO:4, SEQ ID NO:6, and SEQ ID NO:8;
6 c. a peptide comprising at least 6 amino acid residues
7 corresponding to the sequence of SEQ ID NO:2; and

8 d. a peptide comprising at least 6 amino acid residues
9 corresponding to the sequence of SEQ ID NO:4, SEQ ID NO:6, and SEQ ID NO:8.

1 14. The peptide product of claim 13, wherein the peptide is labelled.

1 15. The peptide product of claim 13, wherein the peptide is a fusion
2 protein.

1 16. Use of a peptide as set forth in any one of claims 13 through 15 as
2 an immunogen for the production of antibodies.

1 17. An antibody produced in accordance with claim 16.

1 18. The antibody of claim 17, wherein the antibody is labelled.

1 19. The antibody of claim 17, wherein the antibody is bound to a solid
2 support.

1 20. The antibody of claim 17, wherein the antibody is monoclonal.

1 21. A method to determine the presence or absence of the common
2 hereditary hemochromatosis (HH) gene mutation in an individual comprising:
3 providing DNA or RNA from the individual; and
4 assessing the DNA or RNA for the presence or absence of the HH-
5 associated allele A of a base-pair mutation designated herein 24d1,
6 wherein, as a result, the absence of the allele indicates the absence of the
7 HH gene mutation in the genome of the individual and the presence of the allele indicates
8 the presence of the HH gene mutation in the genome of the individual.

1 22. The method of claim 21, wherein the method further comprises
2 assessing the RNA or DNA for the presence of 24d2.

1 23. The method of claim 21, wherein the method further comprises
 2 assessing the RNA or DNA for the presence of at least one of polymorphisms HHP-1,
 3 HHP-19, or HHP-29, or microsatellite repeat alleles 19D9:205; 18B4:235; 1A2:239;
 4 1E4:271; 24E2:245; 2B8:206; 3321-1:98; 4073-1:182; 4440-1:180; 4440-2:139; 731-
 5 1:177; 5091-1:148; 3216-1:221; 4072-2:170; 950-1:142; 950-2:164; 950-3:165; 950-
 6 4:128; 950-6:151; 950-8:137; 63-1:151; 63-2:113; 63-3:169; 65-1:206; 65-2:159; 68-
 7 1:167; 241-5:108; 241-29:113; 373-8:151; and 373-29:113, D6S258:199, D6S265:122,
 8 D6S105:124; D6S306:238; D6S464:206; and D6S1001:180,

1 24. The method of claim 22, wherein the method further comprises
 2 assessing the RNA or DNA for the presence of at least one of polymorphisms HHP-1,
 3 HHP-19, or HHP-29, or microsatellite repeat alleles 19D9:205; 18B4:235; 1A2:239;
 4 1E4:271; 24E2:245; 2B8:206; 3321-1:98; 4073-1:182; 4440-1:180; 4440-2:139; 731-
 5 1:177; 5091-1:148; 3216-1:221; 4072-2:170; 950-1:142; 950-2:164; 950-3:165; 950-
 6 4:128; 950-6:151; 950-8:137; 63-1:151; 63-2:113; 63-3:169; 65-1:206; 65-2:159; 68-
 7 1:167; 241-5:108; 241-29:113; 373-8:151; and 373-29:113, D6S258:199, D6S265:122,
 8 D6S105:124; D6S306:238; D6S464:206; or D6S1001:18.

1 25. A method for diagnosing whether a patient is afflicted with
 2 hereditary hemochromatosis (HH) disease, comprising:
 3 a. contacting cells of the patient with antibodies directed against
 4 an epitope on an HH protein product corresponding substantially to SEQ ID NO:2 and
 5 b. observing whether the antibodies localize on the cells,
 6 wherein, the observing step, if antibodies do not localize to the cell it is
 7 likely that the patient is afflicted with HH.

1 26. The method of claim 25, wherein the method is conducted *in vitro*.

1 27. The method of claim 25, wherein the method is conducted *in vivo*.

1 28. A method for treating a patient diagnosed as having hereditary
 2 hemochromatosis (HH) disease and homozygous for a 24d1(A) mutation, comprising

3 delivering a polypeptide corresponding to the amino acid sequence of SEQ ID NO:2 to
4 tissues of the patient.

1 29. The method of claim 28, wherein the polypeptide is delivered
2 directly to the tissues.

1 30. The method of claim 28, wherein the polypeptide is delivered
2 intravenously.

1 31. The method of claim 28, wherein the polypeptide is delivered to the
2 tissues through gene therapy.

1 32. An animal model for hereditary hemochromatosis (HH) disease,
2 comprising a mammal possessing a mutant or knocked-out HH gene.

1 33. Metal chelation agents derived from nucleic acid sequences in
2 accordance with claim 1 or from a peptide product in accordance with Claim 13 in a
3 physiologically acceptable carrier.

1 34. The chelation agent of claim 33, wherein the metal is selected from
2 the group consisting of iron, mercury, cadmium, lead, and zinc.

1 35. A method to screen mammals for susceptibility to metal toxicities,
2 comprising, screening such mammals for a mutation in the HH gene and wherein those
3 mammals identified as having a mutation are more susceptible to metal toxicities than
4 mammals not identified as having a mutation.

1 36. The method of claim 35, wherein the metal is selected from the
2 group consisting of iron, mercury, cadmium, lead, and zinc.

1 37. A method for selecting patients infected with hepatitis virus for α -
2 interferon treatment, comprising screening such patients for a mutation in the HH gene

3 and wherein those patients not identified as having a mutation are selected to proceed with
4 α -interferon treatment and those identified as having a mutation are selected to undergo
5 phlebotomy prior to α -interferon treatment.

1 38. A T-cell differentiation factor comprising a moiety selected from the
2 group consisting of molecules derived from nucleic acid sequences in accordance with
3 claim 1 and from a peptide product in accordance with claim 13.

1 39. A method for screening potential therapeutic agents for activity in
2 connection with HH disease, comprising:
3 providing a screening tool selected from the group consisting of a
4 cell line, a cell free extract, and a mammal containing or expressing a defective HH gene
5 or gene product;
6 contacting the screening tool with the potential therapeutic agent;
7 and
8 assaying the screening tool for an activity selected from the group
9 consisting of HH protein folding, iron uptake, iron transport, iron metabolism, receptor-
10 like activities, upstream processes, downstream processes, gene transcription, and
11 signaling events.

1 40. A therapeutic agent for the mitigation of injury due to oxidative
2 processes *in vivo*, comprising a moiety selected from the group consisting of molecules
3 derived from nucleic acid sequences in accordance with claim 1 and from a peptide
4 produce in accordance with claim 13.

1 41. A method for diagnosing a patient as having an increased risk of
2 developing HH disease, comprising:
3 providing DNA or RNA from the individual; and
4 assessing the DNA or RNA for the presence or absence of
5 the HH-associated allele A or a base mutation designated herein 24d1 in combination with
6 assessing the DNA or RNA for the HH-associated allele G of a base mutation designated
7 herein 24d2,

8 wherein, as a result, the absence of the alleles indicates the absence of the
9 HH gene mutation in the genome of the individual and the presence of the alleles
10 indicates the presence of the HH gene mutation in the genome of the individual and an
11 increase risk of developing HH disease.

1 42. The method of claim 41, wherein the method further comprises
2 assessing the RNA or DNA for the presence of at least one of polymorphisms HHP-1,
3 HHP-19, or HHP-29, or microsatellite repeat alleles 19D9:205; 18B4:235; 1A2:239;
4 1E4:271; 24E2:245; 2B8:206; 3321-1:98; 4073-1:182; 4440-1:180; 4440-2:139; 731-
5 1:177; 5091-1:148; 3216-1:221; 4072-2:170; 950-1:142; 950-2:164; 950-3:165; 950-
6 4:128; 950-6:151; 950-8:137; 63-1:151; 63-2:113; 63-3:169; 65-1:206; 65-2:159; 68-
7 1:167; 241-5:108; 241-29:113; 373-8:151; and 373-29:113, D6S258:199, D6S265:122,
8 D6S105:124; D6S306:238; D6S464:206; and D6S1001:180.

1 43. A therapeutic agent for the mitigation of iron overload, comprising
2 a moiety selected from the group consisting of molecules derived from nucleic acid
3 sequences in accordance with claim 1 and from a peptide product in accordance with
4 claim 13.

1 44. A method for treating hereditary hemochromatosis (HH) disease
2 comprising:
3 providing an antibody directed against an HH protein sequence or
4 peptide product; and
5 delivering the antibody to affected tissues or cells in a patient having
6 HH.

1 45. An antisense oligonucleotide directed against a transcriptional
2 product of a nucleic acid sequence selected from the group consisting of SEQ ID NO:1,
3 SEQ ID NO:3, SEQ ID NO:5, SEQ ID NO:7, SEQ ID NO:9, SEQ ID NO:10, SEQ ID
4 NO:11, and SEQ ID NO:12.

1 46. An oligonucleotide of at least 8 nucleotides in length selected from
2 nucleotides 1-46, 48-123; 120-369; 365-394; 390-540; 538-646; 643-1004; 1001-1080;
3 1083-1109; 1106-1304; 1301-1366; 1363-1386; 1389-1514; 1516-1778; 1773-1917; 1921-
4 2010; 2051-2146; 2154-2209; 2234-2368; 2367-2422; 2420-2464; 2465-2491; 2488-2568;
5 2872-2901; 2902-2934; 2936-2954; 2449-3001; 3000-3042; 3420-3435; 3451-3708; 3703-
6 3754; 3750-3770; 3774-3840; 3840-3962; 3964-3978; 3974-3992; 3990-4157; 4153-4251;
7 4257-4282; 4284-4321; 4316-4333; 4337-4391; 4386-4400; 4398-4436; 4444-4547; 4572-
8 4714; 4709-4777; 5165-5397; 5394-6582; 5578-5696; 5691-5709; 5708-5773; 5773-5816;
9 5818-5849; 5889-6045; 6042-6075; 6073-6108; 6113-6133; 6150-6296; 6292-6354; 6356-
10 6555; 6555-6575; 6575-6616; 6620-6792; 6788-6917; 6913-7027; 7023-7061; 7056-7124;
11 7319-7507; 7882-8000; 7998-8072; 8073-8098; 9000-9037; 9486-9502; 9743-9811; 9808-
12 9831; 9829-9866; 9862-9986; 9983-10075; 10072-10091; 10091-10195; 10247-10263;
13 10262-10300; 10299-10448; 10448-10539; 10547-10564; 10580-10612; 10608-10708;
14 10703-10721; 10716-10750; 10749-10774; 10774-10800; and 10796-10825 of SEQ ID
15 NO:1, 3, 5, or 7.

1 47. An oligonucleotide pair comprising an oligonucleotide of claim 46 and
2 an oligonucleotide of at least 8 nucleotides in length selected from SEQ ID NO:1, 3, 5,
3 or 7.

1 48. An oligonucleotide of at least 9 nucleotides in length selected from
2 nucleotides 1-47; 47-124; 119-370; 364-395; 389-541; 537-647; 642-1005; 1000-1081;
3 1082-1110; 1105-1305; 1300-1367; 1362-1387; 1388-1515; 1515-1918; 1920-2011; 2050-
4 2147; 2153-2210; 2233-2369; 2366-2423; 2419-2465; 2464-2492; 2487-2569; 2871-2935;
5 2935-3002; 2999-3043; 3419-3436; 3450-3755; 3749-3771; 3773-3841; 3839-3963; 3963-
6 3979; 3973-3993; 3989-4158; 4152-4252; 4256-4283; 4283-4334; 4336-4401; 4397-4437;
7 4443-4548; 4571-4778; 5164-5398; 5393-5583; 5577-5710; 5707-5774; 5772-5817; 5817-
8 5850; 5888-6046; 6041-6076; 6072-6109; 6112-6134; 6149-6355; 6355-6556; 6554-6576;
9 6574-6793; 6787-7125; 7318-7508; 7881-8001; 7997-8073; 8072-8099; 8999-9038; 9485-
10 9503; 9742-9812; 9807-9832; 9828-9867; 9861-9987; 9982-10076; 10071-10092; 10090-
11 10196; 10246-10264; 10261-10301; 10298-10449; 10447-10540; 10546-10565; 10579-
12 10751; 10748-10775; 10773-10801; and 10795-10825 of SEQ ID NO:1, 3, 5, or 7.

1 49. An oligonucleotide pair comprising an oligonucleotide of claim 48 and
2 an oligonucleotide of at least 8 nucleotides in length selected from SEQ ID NO:1, 3, 5,
3 or 7.

1 50. An oligonucleotide of at least 10 nucleotides in length selected from
2 nucleotides 1-48; 46-125; 118-1006; 999-1082; 1081-1111; 1104-1306; 1299-1368; 1361-
3 1388; 1387-1516; 1514-1919; 1919-2012; 2049-2148; 2152-2211; 2232-2370 2365-2424;
4 2418-2466; 2463-2493; 2486-2570; 2870-2936; 2934-3003; 2998-3044; 3418-3437; 3449-
5 3772; 3772-3842; 3838-3964; 3962-3994; 3988-4284; 4282-4335; 4335-4402; 4396-4438;
6 4442-4549; 4570-4779; 5163-5711; 5706-5775; 5771-5818; 5816-5851; 5867-6047; 6040-
7 6077; 6071-6110; 6111-6135; 6148-6356; 6354-6577; 6573-7126; 7317-7509; 7880-8074;
8 8071-8100; 8998-9039; 9484-9504; 9741-9813; 9806-9833; 9827-9988; 9981-10093;
9 10089-10197; 10245-10265; 10260-10302; 10297-10450; 10446-10541; 10545-10566;
10 10578-10752; 10747-10776; and 10772-10825 of SEQ ID NO:1, 3, 5, or 7.

1 51. An oligonucleotide pair comprising an oligonucleotide of claim 50 and
2 an oligonucleotide of at least 8 nucleotides in length selected from SEQ ID NO:1, 3, 5,
3 or 7.

1 52. An oligonucleotide of at least 11 nucleotides in length selected from
2 nucleotides 1-49; 45-1389; 1386-1517, 1513-1920; 1918-2013; 2048-2149; 2151-2212;
3 2231-2371; 2364-2425; 2417-2467; 2462-2571; 2869-2937; 2933-3004; 2997-3045; 3417-
4 3438; 3448-3773; 3771-3843; 3837-3965; 3961-3995; 3987-4285; 4281-4336; 4334-4403;
5 4395-4439; 4441-4550; 4569-4780; 5162-5712; 5705-5776; 5770-5819; 5815-5852; 5886-
6 6111; 6100-6136; 6147-6357; 6353-6578; 6572-7127; 7316-7510; 7879-8075; 8070-8101;
7 8997-9040; 9483-9505; 9740-10198; 10244-10266; 10257-10303; 10296-10451; 10445-
8 10542; 10544-10567; 10577-10753; 10746-10777; and 10771-10825 of SEQ ID NO:1, 3,
9 5, or 7.

53. An oligonucleotide pair comprising an oligonucleotide of claim 26 and an oligonucleotide of at least 8 nucleotides in length selected from SEQ ID NO:1, 3, 5, or 7.

54. An oligonucleotide of at least 12 nucleotides in length selected from nucleotides 1-50, 44-1390; 1385-1518; 1512-1921; 1917-2014; 2047-2150; 2150-2213; 2230-2372; 2363-2468; 2461-2572; 2868-2938; 2932-3005; 2996-3046; 3416-3439; 3447-3774; 3770-3844; 3836-3966; 3960-4286; 4280-4337; 4333-4440; 4440-4551; 4568-4781; 5161-5713; 5704-5777; 5669-5820; 5814-5853; 5885-6112; 6109-6137; 6146-6358; 6352-6579; 6571-7128; 7315-7511; 7878-8076; 8069-8102; 8996-9041; 9482-9506; 9739-10199; 10243-10267; 10256-10304; 10295-10452; 10444-10543; 10543-10566; 10576-10754; 10745-10778; and 10770-10825 of SEQ ID NO:1, 3, 5, or 7.

55. An oligonucleotide pair comprising an oligonucleotide of claim 54 and an oligonucleotide of at least 8 nucleotides in length selected from SEQ ID NO:1, 3, 5, or 7.

56. An oligonucleotide of at least 13 nucleotides in length selected from nucleotides 1-51; 43-1391; 1384-1519; 1511-1922; 1916-2015; 2046-2151; 2149-2214; 2229-2469; 2460-2573; 2867-2939; 2931-3047; 3415-3440; 3446-3775; 3769-3845; 3835-3967; 3959-4287; 4279-4338; 4332-4441; 4439-4552; 4567-4782; 5160-5778; 5668-5821; 5813-5854; 5884-6113; 6108-6138; 6145-6359; 6351-6580; 6570-7129; 7314-7512; 7877-8077; 8068-8103; 8995-9042; 9481-9507; 9738-10200; 10242-10453; 10443-10544; 10542-10567; 10575-10779; and 10769-10825 of SEQ ID NO:1, 3, 5, or 7.

57. An oligonucleotide pair comprising an oligonucleotide of claim 56 and an oligonucleotide of at least 8 nucleotides in length selected from SEQ ID NO:1, 3, 5, or 7.

58. An oligonucleotide of at least 14 nucleotides in length selected from nucleotides 1-52; 42-1392; 1383-1520; 1510-1923; 1915-2016; 2045-2152; 2148-2215; 2228-2574; 2866-2940; 2930-3048; 3414-3441; 3445-3776; 3768-3968; 3959-4288; 4278-

4 4339; 4331-4442; 4438-4553; 4566-4783; 5159-5822; 5812-5855; 5883-6114; 6107-6139;
5 6144-6360; 6350-6581; 6569-7130; 7313-7513; 7876-8078; 8067-8104; 8994-9043; 9480-
6 9508; 9737-10201; 10241-10454; 10442-10545; 10541-10568; and 10574-10825 of SEQ
7 ID NO:1, 3, 5, or 7.

1 59. An oligonucleotide pair comprising an oligonucleotide of claim 58
2 and an oligonucleotide of at least 8 nucleotides in length selected from SEQ ID NO:1, 3,
3 5, or 7.

1 60. An oligonucleotide of at least 15 nucleotides in length selected from
2 nucleotides 1-53; 41-1393; 1382-1521; 1509-1924; 1914-2017; 2044-2153; 2147-2216;
3 2227-2575; 2865-2942; 2929-3049; 3413-3442; 3444-3777; 3767-3969; 3958-4289; 4277-
4 4340; 4330-4443; 4437-4554; 4565-4784; 5158-5823; 5811-5856; 5882-6115; 6106-6140;
5 6143-6361; 6349-7131; 7312-7514; 7875-8105; 8993-9044; 9479-9509; 9736-10202;
6 10240-10546; 10540-10569; and 10573-10825 of SEQ ID NO:1, 3, 5, or 7.

1 61. An oligonucleotide pair comprising an oligonucleotide of claim 60
2 and an oligonucleotide of at least 8 nucleotides in length selected from SEQ ID NO:1, 3,
3 5, or 7.

1 62. An oligonucleotide of at least 16 nucleotides in length selected from
2 nucleotides 1-1394; 1381-1925; 1913-2018; 2043-2154; 2146-2217; 2226-2576; 2864-
3 3050; 3412-3443; 3443-3778; 3766-4341; 4329-4444; 4436-4555; 4564-4785; 5157-5857;
4 5881-6116; 6105-6141; 6142-7132; 7311-7515; 7874-8106; 8992-9045; 9478-9510; 9735-
5 10203; 10239-10547; 10539-10570; and 10572-10825 of SEQ ID NO:1, 3, 5, or 7.

1 63. An oligonucleotide pair comprising an oligonucleotide of claim 62
2 and an oligonucleotide of at least 8 nucleotides in length selected from SEQ ID NO:1, 3,
3 5, or 7.

1 64. An oligonucleotide of at least 17 nucleotides in length selected from
2 nucleotides 1-1926; 1912-2019; 2042-2155; 2145-2218; 2225-2577; 2863-3051; 3411-

3 3779; 3765-4342; 4329-4445; 4435-4556; 4563-4786; 5156-5858; 5880-6117; 6104-6142;
4 6141-7133; 7310-7516; 7873-8107; 8991-9046; 9477-9511; 9734-10204; 10238-10548;
5 10538-10571; and 10571-10825 of SEQ ID NO:1, 3, 5, or 7.

1 65. An oligonucleotide pair comprising an oligonucleotide of claim 64
2 and an oligonucleotide of at least 8 nucleotides in length selected from SEQ ID NO:1, 3,
3 5, or 7.

1 66. An oligonucleotide of at least 18 nucleotides in length selected from
2 nucleotides 1-2020; 2041-2156; 2144-2219; 2224-2578; 2862-3052; 3410-3780; 3764-
3 4446; 4434-4557; 4562-4787; 5155-5859; 5879-6118; 6103-6143; 6140-7134; 7309-7517;
4 7872-8108; 8990-9047; 9476-9512; 9733-10205; 10237-10549; 10537-10572; and 10570-
5 10825 of SEQ ID NO:1, 3, 5, or 7.

1 67. An oligonucleotide pair comprising an oligonucleotide of claim 66
2 and an oligonucleotide of at least 8 nucleotides in length selected from SEQ ID NO:1, 3,
3 5, or 7.

1 68. An oligonucleotide of at least 8 nucleotides in length selected from
2 nucleotides 1-55; 55-251; 250-306; 310-376; 380-498; 500-528; 516-543; 541-578; 573-
3 592; 590-609; 611-648; 642-660; 664-717; 712-727; 725-763; 772-828; 813-874; 872-
4 928; 913-942; 940-998; 997-1046; 1054-1071; 1076-1116; 1115-1182; 1186-1207; 1440-
5 1483; 1482-1620; 2003-2055; 2057-2107; 2116-2200; and 2453-2469 of SEQ ID NO:9,
6 10, 11 or 12.

1 69. An oligonucleotide pair comprising an oligonucleotide of claim 68
2 and an oligonucleotide of at least 8 nucleotides in length selected from SEQ ID NO:9, 10,
3 11, or 12.

1 70. An oligonucleotide of at least 9 nucleotides in length selected from
2 nucleotides 1-56; 54-252; 249-307; 309-377; 379-499; 499-529; 515-544; 540-579; 572-
3 593; 589-610; 610-649; 641-661; 663-718; 711-728; 724-764; 771-829; 812-875; 871-

4 929; 912-943; 939-999; 996-1047; 1053-1072; 1075-1117; 1114-1183; 1185-1208; 1439-
5 1484; 1481-1629; 2002-2056; 2056-2108; 2115-2201; and 2452-2470 of SEQ ID NO:9,
6 10, 11 or 12.

1 71. An oligonucleotide pair comprising an oligonucleotide of claim 70
2 and an oligonucleotide of at least 8 nucleotides in length selected from SEQ ID NO:9, 10,
3 11, or 12.

1 72. An oligonucleotide of at least 10 nucleotides in length selected from
2 nucleotides 1-57; 53-253; 248-308; 308-378; 378-500; 498-530; 514-545; 539-580; 571-
3 594; 588-611; 609-662; 662-729; 723-765; 770-876; 870-944; 938-1000; 995-1048; 1052-
4 1073; 1074-1118; 1113-1184; 1184-1209; 1438-1485; 1480-1630; 2001-2057; 2055-2109;
5 2114-2202; and 2451-2471 of SEQ ID NO:9, 10, 11 or 12.

1 73. An oligonucleotide pair comprising an oligonucleotide of claim 72
2 and an oligonucleotide of at least 8 nucleotides in length selected from SEQ ID NO:9, 10,
3 11, or 12.

1 74. An oligonucleotide of at least 11 nucleotides in length selected from
2 nucleotides 1-58; 52-254; 247-309; 307-379; 377-501; 497-531; 513-546; 538-595; 587-
3 612; 608-663; 661-730; 722-766; 769-877; 869-1049; 1051-1074; 1073-1119; 1112-1185;
4 1183-1210; 1437-1486; 1479-1631; 2000-2058; 2054-2110; 2113-2203; and 2450-2472 of
5 SEQ ID NO:9, 10, 11 or 12.

1 75. An oligonucleotide pair comprising an oligonucleotide of claim 76
2 and an oligonucleotide of at least 8 nucleotides in length selected from SEQ ID NO:9, 10,
3 11, or 12.

1 76. An oligonucleotide of at least 12 nucleotides in length selected from
2 nucleotides 1-255; 246-310; 306-380; 376-502; 496-596; 586-613; 607-664; 660-767;
3 768-1050; 1050-1075; 072-1120; 1111-1186; 1182-1211; 1436-1487; 1478-1632; 1999-
4 2059; 2053-2121; 2112-2204; and 2449-2473 of SEQ ID NO:9, 10, 11 or 12.

1 77. An oligonucleotide pair comprising an oligonucleotide of claim 76
2 and an oligonucleotide of at least 8 nucleotides in length selected from SEQ ID NO:9, 10,
3 11, or 12.

1 78. An oligonucleotide of at least 13 nucleotides in length selected from
2 nucleotides 1-311; 305-381; 375-503; 495-614; 606-665; 659-768; 767-1051; 1049-1076;
3 1071-1121; 1110-1187; 1181-1212; 1435-1633; 1998-2060; 2052-2205 and 2448-2474 of
4 SEQ ID NO:9, 10, 11 or 12.

1 79. An oligonucleotide pair comprising an oligonucleotide of claim 78
2 and an oligonucleotide of at least 8 nucleotides in length selected from SEQ ID NO:9, 10,
3 11, or 12.

1 80. An oligonucleotide of at least 14 nucleotides in length selected from
2 nucleotides 1-312; 304-382; 374-504; 494-615; 605-666; 658-769; 766-1052; 1048-1077;
3 1070-1188; 1180-1213; 1434-1634; 1997-2061; 2051-2206; and 2447-2475 of SEQ ID
4 NO:9, 10, 11 or 12.

1 81. An oligonucleotide pair comprising an oligonucleotide of claim 80
2 and an oligonucleotide of at least 8 nucleotides in length selected from SEQ ID NO:9, 10,
3 11, or 12.

1 82. An oligonucleotide of at least 15 nucleotides in length selected from
2 nucleotides 1-313; 303-383; 373-505; 493-616; 604-667; 657-770; 765-1053; 1047-1078;
3 1069-1189; 1179-1214; 1433-1635; 1996-2062; 2050-2207; and 2446-2476 of SEQ ID
4 NO:9, 10, 11 or 12.

1 83. An oligonucleotide pair comprising an oligonucleotide of claim 82
2 and an oligonucleotide of at least 8 nucleotides in length selected from SEQ ID NO 9, 10,
3 11, or 12.

1 84. An oligonucleotide of at least 16 nucleotides in length selected from
2 nucleotides 1-314; 302-384; 372-668; 656-771; 764-1054; 1046-1079; 1068-1190; 1178-
3 1215; 1432-1636; 1995-2208; and 2445-2477 of SEQ ID NO:9, 10, 11 or 12.

1 85. An oligonucleotide pair comprising an oligonucleotide of claim 84
2 and an oligonucleotide of at least 8 nucleotides in length selected from SEQ ID NO:9, 10,
3 11, or 12.

1 86. An oligonucleotide of at least 17 nucleotides in length selected from
2 nucleotides 1-315; 301-385; 371-669; 655-772; 763-1055; 1045-1080; 1067-1191; 1177-
3 1216; 1431-1637; 1994-2209; and 2444-2478 of SEQ ID NO:9, 10, 11 or 12.

1 87. An oligonucleotide pair comprising an oligonucleotide of claim 86
2 and an oligonucleotide of at least 8 nucleotides in length selected from SEQ ID NO:9, 10,
3 11, or 12.

1 88. An oligonucleotide of at least 18 nucleotides in length selected from
2 nucleotides 1-773; 762-1056; 1044-1081; 1066-1192; 1176-1217; 1430-1638; 1993-2210;
3 and 2443-2479 of SEQ ID NO:9, 10, 11 or 12.

1 89. An oligonucleotide pair comprising an oligonucleotide of claim 88
2 and an oligonucleotide of at least 8 nucleotides in length selected from SEQ ID NO:9, 10,
3 11, or 12.

1 90. A kit for detection of a polymorphism in the HH gene in a patient
2 sample, the kit comprising at least one oligonucleotide of at least 8 nucleotides in length
3 selected from the group consisting of SEQ ID NOS: 1, 3, 5, 7, 9, 10, 11, or 12, wherein
4 the oligonucleotide is used to amplify a region of HH DNA or RNA in a patient sample.

1 91. The kit of claim 90, further comprising at least a second
2 oligonucleotide selected from the group consisting of SEQ ID NOS: 1, 3, 5, 7, 9, 10, 11,
3 or 12, wherein the first and second oligonucleotides comprise a primer pair.

Add A9)